PARTICIPANT QUESTIONNAIRE

Medical Information

Knowing the precise details of an individual's mutation in an MTM or CNM gene will add to our understanding of these conditions and is likely to be important for developing treatments.

If you have the genetic report yourself (or any other document that includes details of the genetic diagnosis) please upload a copy. Please also enter the name and contact details of the hospital, medical centre or geneticist centre where the genetic test was performed in the field below. In case anything is missing, we can easily obtain the correct document for you from them.

If you do not have the genetic report yourself (or the results of the genetic test are pending), please enter the name and contact details of the hospital, medical centre or geneticist where the test was performed in the field below. We will then contact them and ask for a copy of the report.

If a genetic test has not been performed yet, please contact your doctor, as knowing the details of the mutation is very important for an appropriate treatment.

If a genetic mutation hasn't been confirmed in one of the MTM/CNM genes, you will need to have had a pathological diagnosis of myotubular/centronuclear myopathy via a **muscle biopsy**. Please note that it is quite likely that only those patients with a confirmed genetic mutation will be able to take part in clinical trials to assess potential treatments for these conditions. However those without a confirmed genetic diagnosis may be able to take part in research projects to find new genes for MTM or CNM.

For more help and information on how to be tested, speak with your doctor or geneticist.

Questions with a red asterisk* are mandatory

Genetic Report

5. **Status of the genetic report** *

- I have the genetic report and will upload a copy
- I do not have the genetic report myself but the genetic test results should be available
- The results of the genetic test are pending
- A genetic test was not performed
- A genetic test has been performed but no mutation was found

6. **If a genetic test was performed, please give the name and location of the testing hospital, medical centre, or laboratory:** (free text box)

7. **If you have the genetic report, please upload it here:** (file upload option)

Muscle Biopsy

8. **Has a muscle biopsy been performed?** *

Yes / No / Don't know

9. **If you answered yes, please tell us the name of the hospital where it was performed:** (free text box)

10. **If you answered yes, please tell us the location (city/country) of the hospital where it was performed:** (free text box)

If you have the muscle biopsy report, please upload it here: (file upload option)

Genetic Mutation

11. **Was the myotubular or centronuclear myopathy caused by a mutation in:** *

MTM1 (x-linked myotubular myopathy)

DNM2 (dynamin 2)

BIN1 (amphysin II)

RYR1 (skeletal muscle ryanodine receptor)

TTN (titin)

Don't know

Other (please specify – free text box)

Diagnosis

12. **Diagnosis, according to the specialist:***

Myotubular Myopathy (MTM)

Centronuclear Myopathy (CNM)

Other (please specify – free text box)

Neuromuscular Examination

13. **Please provide the month and year of the most recent neuromuscular examination, if known:** (free text box)

Motor Function

If you are the patient, please answer these questions about yourself.
If you are a parent or guardian registering a child, please answer these questions on their

behalf.

Motor function describes your ability to move your body. Sitting independently means that you can stay in a sitting position for several minutes, without being supported by another person or a stabilising device (such as a chair back, corset or brace).
Walking independently means without being supported by another person or stabilizing device (such as a walking frame, calipers or walking canes).

Questions with a red asterisk* are mandatory

Motor Function

14. **Best motor function ever achieved: ***

Walking without support
Walking with support
Sitting without support
Never able to walk or sit independently
Don't know

15. **From and until what age was this best motor function achieved?** *Please give your answer in months and years, for example: "From 2 years 1 month, until 16 years 2 months"* (free text box)

16. **Current motor function ***

I am able to walk without support
I am able to walk with support
I cannot walk but can sit independently (without support) I
cannot walk or sit independently
Don't know

17. **From what age has this level of motor function been achieved?** *Please give your answer in months and years, for example: "16 years 2 months"* (free text box)

18. **Do you/they currently use a wheelchair? ***

I do not use a wheelchair
I sometimes use a wheelchair, but am able to walk short distances independently I
always use a wheelchair
Don't know

19. **From what age has this level of motor function been achieved?** *Please give your answer in months and years, for example: "2 years, 0 months"* (free text box)

20. **How easily are you/they able to move your/their eyes?** *

This could be full range movement, or some limited movement where you can follow an object with your eyes, even if you can't move your head to look. If you are uncertain, please ask a medical professional such as your physiotherapist or doctor for advice.

I have full range movement of the eyes

I have some limited eye movement

I have no eye movement

Don't know

21. **Have you/they had spinal surgery for scoliosis?** *

Some MTM or CNM patients suffer from weakness in their back muscles which results in a deformation or 'bending' of their spine called scoliosis. In order to stabilise the spine, they often have surgery done. In case such a surgery is planned, but has not yet been performed, please select 'No'

No
Yes

22. **If you answered 'Yes', please provide the age when scoliosis surgery was performed.**

Please give your answer in months and years, for example "16 years and 2 months".

Other Functions

Respiratory function

Ventilation means breathing support from a mechanical ventilation device in the form of either non- invasive ventilation via a face or nose mask, or invasive ventilation via a tracheostomy (an operation to make an incision in the wind-pipe) or endotracheal tube (a breathing tube is inserted into the wind-pipe). Ventilatory support can be used either all day or for just a few hours.

Feeding function

MTM/CNM patients sometimes have trouble eating and swallowing food orally (by mouth) and therefore have to be fed through a feeding tube. A gastric feeding tube (also called a G-tube or a Peg) is one that goes directly into the stomach through an incision in the tummy. A nasal feeding tube (also called nasogastric tube) is one that goes through the nose and down into the stomach.

Heart function

Many neuromuscular patients are routinely required to have regular echocardiograms (ECHO / Sonogram) and electrocardiograms (ECG). Heart problems in myotubular myopathy and centronuclear myopathy patients are very rare, and it would be unusual for these tests to be 'abnormal'. However, it would still be helpful if you could complete the questions.

Questions with a red asterisk* are mandatory.

Respiratory Function

23. **Was ventilation required at birth? ***

Don't know No Yes

24. **What type of ventilation is currently used? ***

Non-Invasive Ventilation (NIV) via a nose or face mask

Invasive ventilation via endotracheal tube or tracheostomy

No ventilation is currently needed

Don't know

25. **If ventilation is used, from what age has it been required?** (Free text box)
Please try to give your answer in years and months, for example "16 years and 2 months"

26. **How many hours is ventilation currently used for? ***

No ventilation is used

Ventilation is sometimes used as therapy, i.e. not regularly

For several hours a day and/or at night

Full time, i.e. 24 hours a day

Don't know

Antibiotics

27. **How many times have antibiotics for chest infections been required over the past 12 months? ***

None required

Between 1 and 3 times

Between 4 and 6 times

More than 6 times

Don't know

Feeding Function

28. **Is a gastric or nasal tube currently used for feeding? ***

No

Yes, a feeding tube with some oral feeding

Yes, a feeding tube only

Don't know

Heart Function

29. **Have you/they ever had an ABNORMAL echocardiogram (ECHO/Sonogram) result?** *

Don't know
No
Yes
No echocardiogram has been done

30. **Have you/they ever had an ABNORMAL electrocardiogram (ECG) result?** *

Don't know
No
Yes
No electrocardiogram has been done

Additional Information

Family information: Since myotubular myopathy and centronuclear myopathy are inherited conditions, it is important for us to know if there are any relatives with similar symptoms or the same diagnosis.

Other Registries: Knowing whether you are registered elsewhere will help us to be accurate when we are estimating the prevalence of these conditions.

Clinical Trials: Knowing whether you have ever taken part in a clinical trial will help us track the development and availability of new therapies and provide information to help with planning and feasibility studies for new clinical trials.

How you found us: This helps us to concentrate our efforts on the best ways to find other people who might want to join the registry.

Newsletter Preferences: Here you can tell us whether you would like to receive newsletters and general updates from us.

Questions with a red asterisk* are mandatory.

Family

31. **Do you know of anybody else in your/the patient's family who has been diagnosed with myotubular myopathy, centronuclear myopathy or similar symptoms? ***

Don't know No Yes

32. **To the best of your knowledge, has anyone in your/their family been married to a cousin or other blood relative? ***

Don't know No Yes

Other Registries

33. **Are your details / this patient's details registered with any other MTM or CNM-related registry or natural history study? ***

Don't know

No, this is the only registry I have joined

Yes, I have joined another MTM and CNM related registry and/or natural history study

34. **If you answered 'Yes', please tell us which ones:**
- Congenital Muscle Disease International Registry (CMDIR)
- International Family Registry for Centronuclear and Myotubular Myopathies (Joshua Frase Foundation)
- Don't know
- Other

35. **If you selected 'Other' please name the registry / registries:** (free text box)

Clinical trials

36. **Have you ever taken part in a clinical trial?**

Yes, I am currently taking part in a clinical trial
Yes, I have previously taken part in a clinical trial
No, I have never taken part in a clinical trial
I don't know

37. **If you answered Yes, please tell us the full name of the clinical trial.**
(Free text)

How you found us

38. **Please tell us how you heard about this registry.**

- My doctor or other healthcare professional, e.g., physiotherapist, genetic counsellor)
- A patient support group (please tell us which one)
- Information included on a genetic test result
- A newsletter (please tell us which one)
- At a conference or other event (please tell us which one)
- Through social media, e.g., a Facebook group (please tell us which one)
- Word of mouth, from friends or family
- Other (please specify)

Newsletter Preferences

39. **Would you like to receive general email communications relevant to Myotubular and Centronuclear Myopathy, such as newsletters, research results and standards of care?***
Yes / No

Thank You for Your Registration

You have now completed the questionnaire.

Thank you for registering your details with the Myotubular and Centronuclear Myopathy Patient Registry.

Please make sure you have completed all the information you can. If there is any important information missing, we may contact you to ask about it.

Remember you can log back in at any time, using your email address and password, to complete or update your details.

You can now log out, or use the 'Modules' menu above to go back to different sections and add more information.

If you have any questions, please contact the Registry Curator at mtmcnmregistry@newcastle.ac.uk for help.

Any further comments?

40. **If you have any feedback or comments on the registration process, please tell us here** (free text box)